GJB3 gene

gap junction protein beta 3

Normal Function

The *GJB3* gene provides instructions for making a protein called gap junction beta 3, more commonly known as connexin 31. This protein is part of the connexin family, a group of proteins that form channels called gap junctions on the surface of cells. Gap junctions open and close to regulate the flow of nutrients, charged atoms (ions), and other signaling molecules from one cell to another. They are essential for direct communication between neighboring cells.

Connexin 31 is found in several different parts of the body, including the outermost layer of the skin (the epidermis) and structures of the inner ear. Connexin 31 plays a role in the growth and maturation of cells in the epidermis. The exact role of this protein in the inner ear is less clear, although it appears to be involved in hearing.

Health Conditions Related to Genetic Changes

erythrokeratodermia variabilis et progressiva

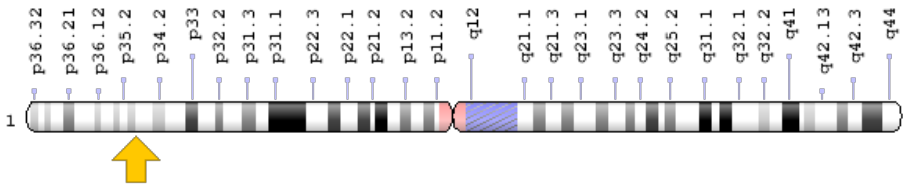
At least 10 *GJB3* gene mutations have been identified in people with erythrokeratodermia variabilis et progressiva (EKVP), a skin disorder characterized by areas of hyperkeratosis, which is abnormally thickened skin, and temporarily reddened patches called erythematous areas. Each of these mutations changes a single protein building block (amino acid) used to make connexin 31. Studies suggest that the abnormal protein can build up in a cell structure called the endoplasmic reticulum (ER), triggering a harmful process known as ER stress. Researchers suspect that ER stress damages and leads to the premature death of cells in the epidermis. This cell death leads to skin inflammation, which appears to underlie the development of erythematous areas. The mechanism by which epidermal damage and cell death contributes to hyperkeratosis is poorly understood.

nonsyndromic hearing loss

Chromosomal Location

Cytogenetic Location: 1p34.3, which is the short (p) arm of chromosome 1 at position 34.3

Molecular Location: base pairs 34,781,189 to 34,786,366 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- connexin 31
- CX31
- CXB3_HUMAN
- DFNA2
- gap junction protein, beta 3, 31kDa
- PNHI

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Gap Junctions Allow Ions and Small Molecules to Flow between Communicating Cells
  https://www.ncbi.nlm.nih.gov/books/NBK22492/
- Madame Curie Bioscience Database: Gap Junctions: Cell-Cell Channels in Animals
  https://www.ncbi.nlm.nih.gov/books/NBK6455/
- Molecular Biology of the Cell (fourth edition, 2002): Gap Junctions Allow Small Molecules to Pass Directly from Cell to Cell
  https://www.ncbi.nlm.nih.gov/books/NBK26857/#A3494

GeneReviews

- Deafness and Hereditary Hearing Loss Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1434

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28GJB3%5BTI%5D%29+OR+%28%28connexin+31%5BTIAB%5D%29+OR+%28CX31%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- GAP JUNCTION PROTEIN, BETA-3
  http://omim.org/entry/603324

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GJB3%5Bgene%5D
- HGNC Gene Family: Deafness associated genes
  http://www.genenames.org/cgi-bin/genefamilies/set/1152
- HGNC Gene Family: Gap junction proteins
  http://www.genenames.org/cgi-bin/genefamilies/set/314
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4285
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2707
- The Connexin-Deafness Homepage
  http://davinci.crg.es/deafness/index.php?seccion=connexins&mut=cx31
- UniProt
  http://www.uniprot.org/uniprot/O75712

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